

Supplementary Data

SUPPLEMENTARY TABLE S1. PHENOTYPE DETAILS OF AFFECTED INDIVIDUALS FROM FAMILY 1

<i>Phenotype</i>	<i>Family1.IV.2 (Proband)</i>	<i>Family1.IV.4 (Brother)</i>	<i>Family1.III.2 (Maternal uncle)</i>
Age (years)	7	3	25
ID	Moderate	Moderate	Moderate
IQ	35	44	Not done
Independent walking	Delayed (2 years)	Delayed (2 years)	Data not available
First words	Delayed (6 years)	Delayed (3 years)	Delayed (9 years)
Seizure	Single, febrile	Single, febrile	None
Head circumference	44 cm; -6 SD	45 cm; -4 SD	54 cm; -1.5 SD
Microcephaly	Yes	Yes	Borderline
Stature	Short (96 cm)	Average (90 cm)	Average/tall
Obesity	No	Yes	No
Penis	Ordinary	Micropenis	Ordinary
Testes	Ordinary	Hypoplastic	Ordinary
Face	Elongated	Elongated	Low
Anterior hairline	Ordinary	Low	Low
Ear position	Low set	Low set	Low set
Eyebrows	Bushy	Bushy	Bushy
Synophrys	Yes	Yes	Yes
Philtrum	Short	Ordinary	Short
Palate	High arched	Ordinary	Ordinary
Teeth	Normal	Spaced	Spaced
Long and/or prominent ears	Yes	Yes	Yes
Broad ear lobule	Yes	Yes	Yes
Neck	Short	Short	Short
Chest	Ordinary	Ordinary	Mild pectus excavatum
Simian crease	No	Unilateral (left)	No
Pes planus (flat feet)	Yes	Yes	Yes
Pigmentary skin change/dry skin	No	No	Yes
Posture	Normal	Normal	Stooping
Behavior	ADHD	ADHD	Aggression

ADHD, attention-deficit hyperactivity disorder.